ORGANIC ACID DISORDERS

What is it?

Organic acid disorders are a group of inherited metabolic conditions. Each organic acid disorder is associated with a specific enzyme deficiency that causes the accumulation of organic acids in the blood and urine. The accumulated compounds or their metabolites, are toxic, resulting in the clinical features of these disorders.

How do you get it?

All organic acid disorders are inherited in an autosomal recessive pattern. As an autosomal recessive disorder, the parents of a child with one of these conditions are unaffected, healthy carriers of the condition, and have one normal gene and one abnormal gene. With each pregnancy, carrier parents have a 25 percent chance of having a child with two copies of the abnormal gene and the resulting organic and acid defect. Carrier parents have a 50 percent chance of having an unaffected, non-carrier child. These risks would hold true of each pregnancy. All siblings of infants diagnosed with an organic acid disorder should be tested.

Many organic acid disorders present in the neonatal period. Typically, an affected newborn appears normal for the first days of life, but then may develop vomiting, poor feeding, failure to thrive, hypoglycemia, hyperammonemia, seizures, hypotonia and lethargy, progressing to coma. Common features include ketotic hyperglycinemia, metabolic acidosis, and in some cases an unusual odor. Many individuals affected with organic acid disorders have a significant risk of dying during infancy.

How common is it?

Estimates vary widely for the incidence of each organic acid disorder and for many the actual incidence is not yet known, however, most of these disorders are rare.

How is it treated?

For many organic acid disorders, early diagnosis and treatment can significantly change the outcome of the disease. Treatment depends upon the specific organic acid disorder.

If the child needs additional testing or diagnostic evaluation it is important that you follow through. Treatment is life long and compliance with dietary management is imperative to your child’s health, growth and development.

Infants and children with an organic acid disorder should have regular follow-up appointments with a metabolic disease specialist.
If your infant shows early signs of the condition, such as vomiting or lethargy, immediately seek medical care. The metabolic specialist and the primary care provider should develop a medical plan for these acute episodes.

Long-term management, monitoring and compliance with treatment recommendations are essential to your child's well being. A multi-disciplinary approach, including the following specialties is recommended: pediatrics, genetics and nutrition.

Where can I get services?

Cardinal Glennon Memorial Hospital for Children
St. Louis, MO
314-577-5639

Children's Mercy Hospital
Kansas City, MO
816-234-3804

St. Louis Children's Hospital
St. Louis, MO
314-454-6051

University Hospital and Clinics
Columbia, MO
573-882-6979

Related Links

Centers for Disease Control and Prevention www.cdc.gov

Medline Plus (National Library of Medicine and the National Institutes of Health) www.medlineplus.gov

National Coalition for PKU and Allied Disorders
www.pku-allieddisorders.org

National Library of Medicine (NLM) Pub Med www.ncbi.nlm.nih.gov

Organic Acid Association www.oaanews.org